Advancing breakthrough science into **life-changing medicines for rare disease**

A bold scientific initiative is advancing promising rare disease discoveries, aiming to turn breakthrough research into future treatments for patients.

Cindy (left) and Noelle ("Noni") Schulz | Photo courtesy of Cindy Schulz

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Two years ago, the Oxford-Harrington Rare Disease Centre (OHC) made a commitment to advance 40 rare disease drugs into clinical trials by 2034. The partnership already has over 50 drugs in development — reflecting the urgency of unmet need and the strength of its model.

Central to this effort is its Rare Disease Scholar Award programme. Each year, ten academic investigators from the US, UK and Canada are selected to receive funding and translational support on projects ranging from neurodevelopmental, neuromuscular and metabolic disorders to rare cancers. Uniquely, Scholars also receive strategic guidance on all aspects of drug development, with the goal of accelerating projects towards clinical trials and de-risking them for further development and investment.

Providing hope for families

One of the newest Scholar projects focuses on CASK, a rare genetic disease that affects normal brain development, leading to severe physical and intellectual impairment and other debilitating symptoms. For families, the impact is devastating.

When Cindy Schulz's daughter Noelle ('Noni') was a toddler, developmental delays became impossible to ignore. Despite multiple appointments, the cause of Noni's condition could not be identified, and Cindy was told that there was no treatment plan beyond supportive care.

Noni faced significant intellectual and physical challenges and, although she learned to walk and talk, many everyday skills remained out of reach. So, Cindy focused on building a joyful, fulfilling life for her daughter. Now 42, Noni lives happily with her parents and has enjoyed working at a local grocery store for nearly 20 years.

Noni was 30 when genetic testing finally identified the cause: a rare mutation in the CASK gene. At the time, only around 50 individuals had been diagnosed. Today, CASK is estimated to affect about 2,000 individuals, and that number is growing as awareness and testing improve.

For many families, the hardest part of a rare disease diagnosis is the absence of a clear treatment path while promising academic discoveries often stall before becoming therapies, because of a lack of commercial incentives.

Transforming scientific discovery into treatments

A first-of-its-kind partnership between the University of Oxford and Harrington Discovery Institute at University Hospitals in Cleveland, the OHC connects world-class academic science with drug development and industry expertise to help transform promising discoveries into therapies and cures.

Enter Dr Mingshan Xue, a neuroscientist at Baylor College of Medicine. Dr Xue has long studied the genetic pathways that shape brain development, and his interest in CASK began when he met a patient during his postdoctoral training. Confronted by its severity and the lack of effective treatments, he committed to finding a therapy. Through connections with families and advocacy groups, such as Project CASK, he understood the urgency.

As a supported Scholar, Dr Xue's research focuses on developing a gene therapy to replace the dysfunctional gene with a functional copy, thereby addressing the root cause of the disease rather than just managing symptoms.

"What attracted me to this unique programme was access to deep drug development expertise," Dr Xue explains. "Gene therapy requires coordinated scientific, regulatory and manufacturing planning. That integrated support is essential to move this toward patients."

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Advancing gene therapy for CASK

If successful, a disease-modifying therapy for CASK could significantly improve the development, health and quality of life of affected individuals.

"When Noni was young, effective therapy wasn't even part of the conversation," Cindy reflects. "Now, researchers are working to change the course of CASK, raising hopes for a cure that families have been praying for. It's extraordinary."

Progress in finding new treatments and cures for CASK and for many other rare diseases that collectively affect nearly 500 million people worldwide[1] will take time, and not every programme will succeed. But the convergence of determined families, focused science and translational partnerships like the OHC is reshaping what is possible.

References:
1. Harrington Discovery Institute. (2025). Oxford-Harrington Rare Disease Centre Announces 2025 Scholars Advancing Promising Treatments.

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